

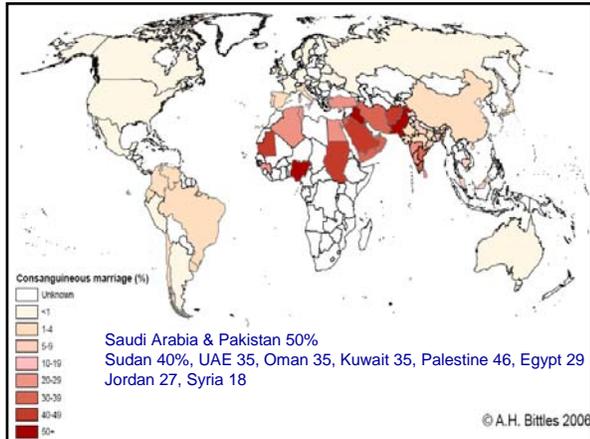
Newborn Screening In Saudi Arabia Country Report



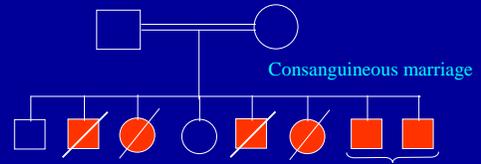
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Population: 22,000,000 (16 M Saudi, 6M expatriates)
450,000 birth per year
Large family size: between 6-12



A Family with Propionic Acidemia



- Affire
- Developmental delay
- Recurrent admission

Screening Milestones for Saudi Arabia

- 1991 Cord blood Hypothyroidism screening at 20 centers
- 1994 Introduction of tandem mass spectrometry at KFSH&RC
- 1995-1998 First MS/MS pilot study, published 1999
- 1995-2005 Expanded newborn screening on small scale
- Aug 2005 National newborn screening started
- Phase I includes 24 birth centers and 120,000 newborns
- 2008 Coverage of >400,00 newborns

HRH Prince Sultan Bin Salman with Excellency Minister of Health (2004)





SNBS Phase I



- ❖ 120,000 newborn blood samples collected in first year
- ❖ 24 hospitals are included from different regions
- ❖ Screen for 16 diseases (Panel A & Panel B)
- ❖ Study logistical and coordination problems
- ❖ Outcome will lead to national registry (still pending)
- ❖ Coordinate recall and tracking problems (still a big problem)

Diseases Covered by the Saudi Newborn Screening

Panel A: Diseases Covered by Tandem Mass Spectrometry:

1. Phenylketonuria (PKU)
2. Maple Syrup Urine Disease (MSUD)
3. Argininosuccinase Deficiency (ASL)
4. Citrullinemia (ASD)
5. HMG-CoA Lyase Deficiency (HMG)
6. Isovaleric Acidemia (IVA)
7. Methylmalonic Acidemia (MMA)
8. Propionic Acidemia (PA)
9. Beta-keto Thiolase Deficiency (BKT)
10. Glutaric Acidemia type-I (GA-I)
11. Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
12. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)

Panel B: Diseases Covered by Kit-based Assays:

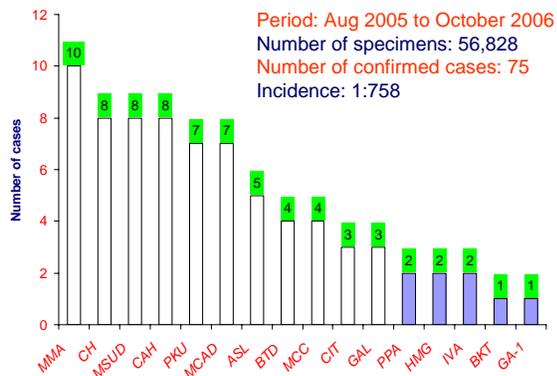
1. Galactosemia
2. Congenital Hypothyroidism (CH)
3. Congenital Adrenal Hyperplasia (CAH)
4. Biotinidase Deficiency (BD)



Technology

- 4 Tandem mass spectrometry (MS/MS & LC-MS/MS).
- 2 Gas chromatography mass spectrometry (GC-MS)
- 4 High pressure liquid chromatography (HPLC)
- 1 UPLC system
- 1 Autodelfia TSH & CAH
- 1 VictorD2 BTD & GALAC
- 3 Blood spots punchers
- 1 Lab Database since 1994
- Relatively small lab space

Scientific & Technical Staff



Recall is a nightmare

3 months Sample

	Questionable MS/MS	Recalled	Failed Recall
	90	10	80
		11%	89%
	Questionable 17-OHP	Recalled	Failed Recall
	217	28	189
		13%	87%
	Questionable TSH	Recalled	Failed Recall
	167	7	160
		4%	96%
	Questionable BTD	Recalled	Failed Recall
	54	2	52
		4%	96%
	Questionable Galt	Recalled	Failed Recall
	40	1	39
		2.5%	97.5%

Barriers to Improving SNBS

- *Incomplete or incorrect demographics*
- *Early specimen collection*
- *Communications and resource difficulties*
- *Recall difficulties*
- *Inadequate oversight & tracking & medications*
- *Deficiency in metabolists, dietitians, genetic counselors*
- *Uncertainty about financing & politics*
- *A significant single improvement "Mandating NBS by Law"*

Thank you